

Expression of *HAL* in skin

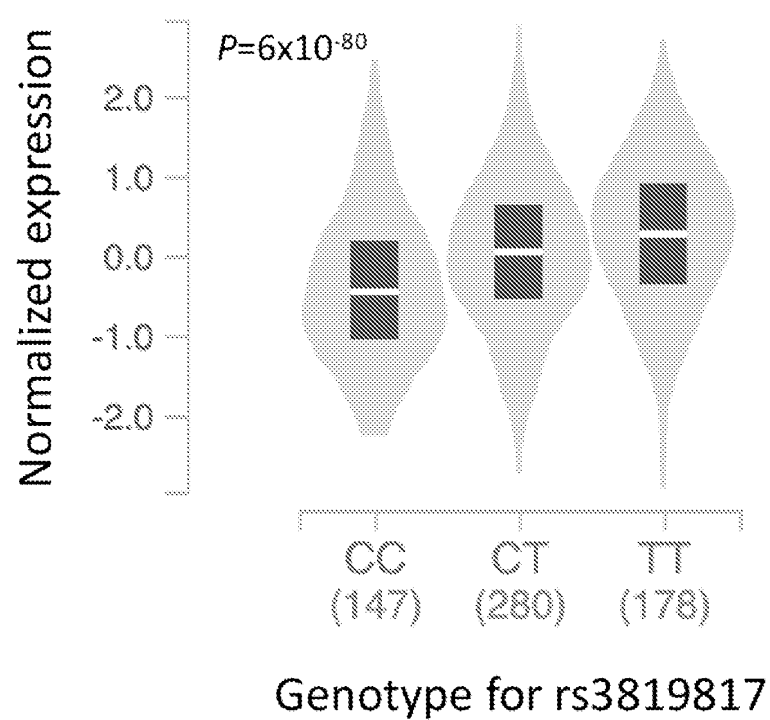


Figure 1

Phenotype	Study	Effect [95% CI]	P-value	N cases with 0 1 2 copies of the effect allele	N controls with 0 1 2 copies of the effect allele	EAF
<i>Burden of rare pLOF and deleterious missense variants in HAL</i>						
Vitamin D	UKB	0.116[0.099,0.132]	3.70E-43	381231 13124 27	NA NA NA	0.0167
	GHS	0.059[0.017,0.100]	5.80E-03	54242 2039 1	NA NA NA	0.0181
Skin tanning	Meta-analysis	0.108[0.093,0.123]	1.80E-43	435473 15163 28	NA NA NA	0.0169
	UKB	1.138[1.091,1.187]	2.20E-09	86331 13244 9	322123 10845 18	0.0167
Actinic keratosis	UKB	1.230[1.020,1.483]	3.00E-02	2887 123 0	371288 12749 25	0.0167
	GHS	1.244[1.106,1.399]	2.80E-04	9758 426 0	103672 3874 2	0.0183
Non melanoma skin cancer	Meta-analysis	1.240[1.122,1.370]	2.40E-05	12645 549 0	474960 16623 27	0.0171
	UKB	1.136[1.062,1.216]	2.10E-04	26054 996 4	388311 13299 23	0.0167
	GHS	1.039[0.891,1.210]	6.30E-01	55601 208 0	105832 3998 2	0.0182
	Meta-analysis	1.120[1.052,1.191]	3.40E-04	31614 1204 4	494143 17297 25	0.017
Melanoma skin cancer	UKB	1.123[0.944,1.336]	1.90E-01	3863 148 0	411891 14203 27	0.0167
	GHS	1.178[0.872,1.592]	2.90E-01	1203 53 0	109815 4161 3	0.0183
Skin color (dark olive)	Meta-analysis	1.136[0.978,1.321]	9.60E-02	5066 201 0	521706 18364 30	0.0171
	UKB	1.071[0.939,1.222]	3.10E-01	6956 254 0	404178 13946 27	0.0168
<i>GIWAS sentinel variant (rs10859995;C) that is in high LD with a sentinel eQTL (rs3819817;T) that increases HAL expression in skin tissue</i>						
Vitamin D	UKB	-0.040[-0.044,-0.035]	3.60E-74	69201 192291 132890	NA NA NA	0.5808
	GHS	-0.026[-0.037,-0.015]	3.80E-06	11106 27942 17234	NA NA NA	0.5532
Skin tanning	Meta-analysis	-0.038[-0.042,-0.034]	9.00E-78	80307 220233 150124	NA NA NA	0.5775
	UKB	0.954[0.943,0.964]	8.80E-17	16507 44025 29052	57476 162040 113470	0.5811
Actinic keratosis	UKB	0.988[0.947,1.029]	5.50E-01	436 1113 805	80368 224007 155210	0.5814
	GHS	0.941[0.911,0.972]	2.60E-04	2117 5035 3032	21204 53211 33133	0.5533
Non melanoma skin cancer	Meta-analysis	0.951[0.924,0.978]	4.90E-04	2553 6148 3837	101572 277218 188343	0.576
	UKB	0.958[0.941,0.976]	3.40E-06	4995 13075 8958	69988 195779 135576	0.5812
	GHS	0.949[0.912,0.987]	9.40E-03	1295 2905 1829	22436 56179 34883	0.5531
	Meta-analysis	0.956[0.941,0.972]	1.10E-07	6290 15980 10787	92424 251958 170459	0.5753
Melanoma skin cancer	UKB	0.915[0.875,0.957]	9.80E-05	773 1981 1256	74478 207523 143801	0.5812
	GHS	0.928[0.857,1.004]	6.40E-02	2801656 374	23311 58371 36150	0.5531
Skin color (dark olive)	Meta-analysis	0.918[0.883,0.955]	1.70E-05	1053 2637 1630	97789 265894 179951	0.5754
	UKB	0.971[0.939,1.005]	9.30E-02	1322 3567 2321	73177 203844 141130	0.5811

Figure 2